

SEQUENCE LISTING

<110> DREYFUSS, Gideon
CHARROUX, Bernard
PELLIZZONI, Livio
FISCHER, Utz
Liu, Qing

<120> COMPOSITIONS, METHODS AND KITS USEFUL FOR THE DIAGNOSIS AND TREATMENT OF SPINAL MUSCULAR ATROPHY

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<140> US 09/399,081
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<170> PatentIn version 3.1

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 catggcaaga atattatgct gccgcttctc attcatatta ttggaatgct cagagacatc 2700
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 accagtgatt ataggatata cctgagacca tcaggaactg tcaacaaatg ataccttgg 2820
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 tagacttggt tagaaaaaaaaaaaaaa aa 3152

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 <211> 824
 <212> PRT
 <213> Homo sapiens

<400> 8

Met Ala Ala Ala Phe Glu Ala Ser Gly Ala Leu Ala Ala Val Ala Thr
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Ala Met Pro Ala Glu His Val Ala Val Gln Val Pro Ala Pro Glu Pro
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Thr Pro Gly Pro Val Arg Ile Leu Arg Thr Ala Gln Asp Leu Ser Ser
 35 40 45

Pro Arg Thr Arg Thr Gly Asp Val Leu Leu Ala Glu Pro Ala Asp Phe
 50 55 60

Glu Ser Leu Leu Leu Ser Arg Pro Val Leu Glu Gly Leu Arg Ala Ala
 65 70 75 80

Gly	Phe	Glu	Arg	Pro	Ser	Pro	Val	Gln	Leu	Lys	Ala	Ile	Pro	Leu	Gly	
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Arg	Cys	Gly	Leu	Asp	Leu	Ile	Val	Gln	Ala	Lys	Ser	Gly	Thr	Gly	Lys	
100							105					110				
Thr	Cys	Val	Phe	Ser	Thr	Ile	Ala	Leu	Asp	Ser	Leu	Val	Leu	Glu	Asn	
115							120					125				
Leu	Ser	Thr	Gln	Ile	Leu	Ile	Leu	Ala	Pro	Thr	Arg	Glu	Ile	Ala	Val	
130							135				140					
Gln	Ile	His	Ser	Val	Ile	Thr	Ala	Ile	Gly	Ile	Lys	Met	Glu	Gly	Leu	
145							150			155			160			
Glu	Cys	His	Val	Phe	Ile	Gly	Gly	Thr	Pro	Leu	Ser	Gln	Asp	Lys	Thr	
165							170					175				
Arg	Leu	Lys	Lys	Cys	His	Ile	Ala	Val	Gly	Ser	Pro	Gly	Arg	Ile	Lys	
180							185					190				
Gln	Leu	Ile	Glu	Leu	Asp	Tyr	Leu	Asn	Pro	Gly	Ser	Ile	Arg	Leu	Phe	
195							200					205				
Ile	Leu	Asp	Glu	Ala	Asp	Lys	Leu	Leu	Glu	Glu	Ser	Phe	Gln	Glu		
210							215					220				
Gln	Ile	Asn	Trp	Ile	Tyr	Ser	Ser	Leu	Pro	Ala	Ser	Lys	Gln	Met	Leu	
225							230				235			240		
Ala	Val	Ser	Ala	Thr	Tyr	Pro	Glu	Phe	Leu	Ala	Asn	Ala	Leu	Thr	Lys	
245							250					255				
Tyr	Met	Arg	Asp	Pro	Thr	Phe	Val	Arg	Leu	Asn	Ser	Ser	Asp	Pro	Ser	
260							265					270				
Leu	Ile	Gly	Leu	Lys	Gln	Tyr	Tyr	Lys	Val	Val	Asn	Ser	Tyr	Pro	Leu	
275							280					285				
Ala	His	Lys	Val	Phe	Glu	Glu	Lys	Thr	Gln	His	Leu	Gln	Glu	Leu	Phe	
290							295					300				
Ser	Arg	Ile	Pro	Phe	Asn	Gln	Ala	Leu	Val	Phe	Ser	Asn	Leu	His	Ser	
305							310				315			320		
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325							330					335				
Glu	Cys	Ile	Ser	Gly	Asn	Met	Asn	Gln	Asn	Gln	Arg	Leu	Asp	Ala	Met	
340							345					350				
Ala	Lys	Leu	Lys	His	Phe	His	Cys	Arg	Val	Leu	Ile	Ser	Thr	Asp	Leu	
355							360					365				
Thr	Ser	Arg	Gly	Ile	Asp	Ala	Glu	Lys	Val	Asn	Leu	Val	Val	Asn	Leu	
370							375					380				

Asp Val Pro Leu Asp Trp Glu Thr Tyr Met His Arg Ile Gly Arg Ala
 385 390 395 400
 Gly Arg Phe Gly Thr Leu Gly Leu Thr Val Thr Tyr Cys Cys Arg Gly
 405 410 415
 Glu Glu Glu Asn Met Met Arg Ile Ala Gln Lys Cys Asn Ile Asn
 420 425 430
 Leu Leu Pro Leu Pro Asp Pro Ile Pro Ser Gly Leu Met Glu Glu Cys
 435 440 445
 Val Asp Trp Asp Val Glu Val Lys Ala Ala Val His Thr Tyr Gly Ile
 450 455 460
 Ala Ser Val Pro Asn Gln Pro Leu Lys Lys Gln Ile Gln Lys Ile Glu
 465 470 475 480
 Arg Thr Leu Gln Ile Gln Lys Ala His Gly Asp His Met Ala Ser Ser
 485 490 495
 Arg Asn Asn Ser Val Ser Gly Leu Ser Val Lys Ser Lys Asn Asn Thr
 500 505 510
 Lys Gln Lys Leu Pro Val Lys Ser His Ser Glu Cys Gly Ile Ile Glu
 515 520 525
 Lys Ala Thr Ser Pro Lys Glu Leu Gly Cys Asp Arg Gln Ser Glu Glu
 530 535 540
 Gln Met Lys Asn Ser Val Gln Thr Pro Val Glu Asn Ser Thr Asn Ser
 545 550 555 560
 Gln His Gln Val Lys Glu Ala Leu Pro Val Ser Leu Pro Gln Ile Pro
 565 570 575
 Cys Leu Ser Ser Phe Lys Ile His Gln Pro Tyr Thr Leu Thr Phe Ala
 580 585 590
 Glu Leu Val Glu Asp Tyr Glu His Tyr Ile Lys Glu Gly Leu Glu Lys
 595 600 605
 Pro Val Glu Ile Ile Arg His Tyr Thr Gly Pro Gly Asp Gln Thr Val
 610 615 620
 Asn Pro Gln Asn Gly Phe Val Arg Asn Lys Val Ile Glu Gln Lys Val
 625 630 635 640
 Pro Val Leu Ala Ser Ser Ser Gln Ser Gly Asp Ser Glu Ser Asp Ser
 645 650 655
 Asp Ser Tyr Ser Ser Arg Thr Ser Ser Gln Ser Lys Gly Asn Lys Ser
 660 665 670
 Tyr Leu Glu Ser Ser Ser Asp Asn Gln Leu Lys Asp Ser Glu Ser Thr
 675 680 685

Pro Val Asp Asp Arg Ile Ser Leu Glu Gln Pro Pro Asn Gly Thr Asp
690 695 700

Thr Pro Asn Pro Glu Lys Tyr Gln Glu Ser Pro Gly Ile Gln Met Lys
705 710 715 720

Thr Arg Leu Lys Glu Gly Ala Ser Gln Arg Ala Lys Gln Ser Arg Arg
725 730 735

Asn Leu Pro Arg Arg Ser Ser Phe Arg Leu Gln Thr Glu Ala Gln Glu
740 745 750

Asp Asp Trp Tyr Asp Cys His Arg Glu Ile Arg Leu Ser Phe Ser Asp
755 760 765

Thr Tyr Gln Asp Tyr Glu Glu Tyr Trp Arg Ala Tyr Tyr Arg Ala Trp
770 775 780

Gln Glu Tyr Tyr Ala Ala Ala Ser His Ser Tyr Tyr Trp Asn Ala Gln
785 790 795 800

Arg His Pro Ser Trp Met Ala Ala Tyr His Met Asn Thr Ile Tyr Leu
805 810 815

Gln Glu Met Met His Ser Asn Gln
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<210> 9

<211> 797

<212> DNA

<213> Gallus gallus

<400> 9

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taaagaacgg ggactgctca gagccttcgg acaaacagga gcagcggcg ggggtgaaaa 180

ggaaaaacag caagaagaac aggaacagaa acaagagcaa cgccgtgccg ttgaagcagt 240

ggaaagttgg cgacagctgt aacgctgttt ggtctgagga tggtaatgtc taccctgcaa 300

ctattgcctc cataaatctg aagagggta catgcgttgt tacttacacc ggatatggaa 360

acaaggagga acagaacctg gctgatctac ttcctccagc tagcgatgaa acaaatgaaa 420

atgagactcc gtattcaaca gatgaaagtg aaaaatcttc ccagtcacat cacaatgaaa 480

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ctccaggatt gggaggcat ggttcaaat tcagaacact tccaccattc ttgtcttgct 600

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<210> 10
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		20						25					30		

Val	Ala	Ser	Phe	Lys	Asn	Ala	Leu	Lys	Asn	Gly	Asp	Cys	Ser	Glu	Pro
		35						40				45			

Ser	Asp	Lys	Gln	Glu	Gln	Arg	Ala	Gly	Val	Lys	Arg	Lys	Asn	Ser	Lys
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Lys	Asn	Arg	Asn	Arg	Asn	Lys	Ser	Asn	Ala	Val	Pro	Leu	Lys	Gln	Trp
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Lys	Val	Gly	Asp	Ser	Cys	Asn	Ala	Val	Trp	Ser	Glu	Asp	Gly	Asn	Val
		85							90			95			

Tyr	Pro	Ala	Thr	Ile	Ala	Ser	Ile	Asn	Leu	Lys	Arg	Gly	Thr	Cys	Val
		100						105				110			

Val	Thr	Tyr	Thr	Gly	Tyr	Gly	Asn	Lys	Glu	Glu	Gln	Asn	Leu	Ala	Asp
		115						120				125			

Leu	Leu	Pro	Pro	Ala	Ser	Asp	Glu	Thr	Asn	Glu	Asn	Glu	Thr	Pro	Tyr
		130					135				140				

Ser	Thr	Asp	Glu	Ser	Glu	Lys	Ser	Ser	Gln	Ser	His	His	Asn	Glu	Asn
		145				150				155			160		

Asn	Cys	Thr	Lys	Ala	Arg	Phe	Ser	Pro	Lys	Asn	Leu	Arg	Phe	Pro	Ile
		165							170			175			

Pro	Pro	Thr	Pro	Pro	Gly	Leu	Gly	Arg	His	Gly	Ser	Lys	Phe	Arg	Thr
		180						185				190			

Leu	Pro	Pro	Phe	Leu	Ser	Cys	Trp	Pro	Pro	Phe	Pro	Ala	Gly	Pro	
		195						200				205			

Pro	Leu	Ile	Pro	Pro	Pro	Pro	Met	Gly	Pro	Asp	Ser	Pro	Glu	Asp	
		210					215				220				

Asp	Glu	Ala	Leu	Gly	Ser	Met	Leu	Ile	Ala	Trp	Tyr	Met	Ser	Gly	Tyr
		225						230			235		240		

His	Thr	Gly	Tyr	Tyr	Leu	Gly	Leu	Lys	Gln	Ser	Arg	Met	Glu	Ala	Ala
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Leu	Glu	Arg	Glu	Ala	Tyr	Leu	Lys								
				260											
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Gly	Met	Asp	Pro	Asp	Gly	Val	Ile	Glu	Ser	Asn	Trp	Asn	Glu	Ile	Val
					20			25							30
Asp	Asn	Phe	Asp	Asp	Met	Asn	Leu	Lys	Glu	Ser	Leu	Leu	Arg	Gly	Ile
					35			40							45
Tyr	Ala	Tyr	Gly	Phe	Glu	Lys	Pro	Ser	Ala	Ile	Gln	Gln	Arg	Ala	Ile
					50			55							60
Ile	Pro	Cys	Ile	Lys	Gly	Tyr	Asp	Val	Ile	Ala	Gln	Ala	Gln	Ser	Gly
				65			70			75					80
Thr	Gly	Lys	Thr	Ala	Thr	Phe	Ala	Ile	Ser	Ile	Leu	Gln	Gln	Leu	Glu
					85				90						95
Ile	Glu	Phe	Lys	Glu	Thr	Gln	Ala	Leu	Val	Leu	Ala	Pro	Thr	Arg	Glu
					100			105							110
Leu	Ala	Gln	Gln	Ile	Gln	Lys	Val	Ile	Leu	Ala	Leu	Gly	Asp	Tyr	Met
					115				120						125
Gly	Ala	Thr	Cys	His	Ala	Cys	Ile	Gly	Gly	Thr	Asn	Val	Arg	Asn	Glu
					130			135							140
Met	Gln	Lys	Leu	Gln	Ala	Glu	Ala	Pro	His	Ile	Val	Val	Gly	Thr	Pro
					145			150			155				160
Gly	Arg	Val	Phe	Asp	Met	Leu	Asn	Arg	Arg	Tyr	Leu	Ser	Pro	Lys	Trp
					165				170						175
Ile	Lys	Met	Phe	Val	Leu	Asp	Glu	Ala	Asp	Glu	Met	Leu	Ser	Arg	Gly
					180			185							190
Phe	Lys	Asp	Gln	Ile	Tyr	Glu	Ile	Phe	Gln	Lys	Leu	Asn	Thr	Ser	Ile
					195				200						205
Gln	Val	Val	Leu	Leu	Ser	Ala	Thr	Met	Pro	Thr	Asp	Val	Leu	Glu	Val
					210			215							220
Thr	Lys	Lys	Phe	Met	Arg	Asp	Pro	Ile	Arg	Ile	Leu	Val	Lys	Lys	Glu
					225			230			235				240

Glu Leu Thr Leu Glu Gly Ile Lys Gln Phe Tyr Ile Asn Val Glu Arg
245 250 255

Glu Glu Trp Lys Leu Asp Thr Leu Cys Asp Leu Tyr Glu Thr Leu Thr
260 265 270

Ile Thr Gln Ala Val Ile Phe Leu Asn Thr Arg Arg Lys Val Asp Trp
275 280 285

Leu Thr Glu Lys Met His Ala Arg Asp Phe Thr Val Ser Ala Leu His
290 295 300

Gly Asp Met Asp Gln Lys Glu Arg Asp Val Ile Met Arg Glu Phe Arg
305 310 315 320

Ser Gly Ser Ser Arg Val Leu Ile Thr Thr Asp Leu Leu Ala Arg Gly
325 330 335

Ile Asp Val Gln Gln Val Ser Leu Val Ile Asn Tyr Asp Leu Pro Thr
340 345 350

Asn Arg Glu Asn Tyr Ile His Arg Ile Gly Arg Gly Gly Arg Phe Gly
355 360 365

Arg Lys Gly Val Ala Ile Asn Phe Val Thr Glu Glu Asp Lys Arg Ile
370 375 380

Leu Arg Asp Ile Glu Thr Phe Tyr Asn Thr Thr Val Glu Glu Met Pro
385 390 395 400

Met Asn Val Ala Asp Leu Ile
405